21. (Once Amended) A kit for the *in vitro* detection of a defect in the survival motor neuron gene, comprising:

a set of primers, wherein at least one of said primers is contained in the sequence of nucleotides 921 to 1469 of SEQ ID No: 12;

reagents for an amplification reaction; and

a probe for the detection of the amplified product.

23. (Twice Amended) The kit of Claim 53, for the detection of Spinal Muscular Atrophy (SMA).

30. (Once Amended) A method for detecting a defect in the Survival Motor Neuron gene, said method comprising :

(a) extracting DNA from a patient sample;

(b) amplifying said DNA with primers, wherein at least one of said primers is contained in the sequence of nucleotides 921 to 1469 of SEQ ID No: 12;

(c) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP); and

(d) detecting the presence or absence of said defect in the Survival Motor Neuron gene, wherein the presence of said defect is indicative of a Survival Motor Neuron disorder.

31. (Once Amended) The method of claim 30, wherein said detection of a defect in the Survival Motor Neuron gene is indicative of a Spinal Muscular Atrophy.

33. (Once Amended) A method for detecting Spinal Muscular Atrophy, said method comprising:

(a) extracting DNA from a patient sample;

(b) hybridizing said DNA with a DNA probe comprising all or part of the DNA sequence of SEQ ID Nos: 12 or 13 under stringent conditions;

(c) detecting the hybrids formed; and

(d) detecting the presence or absence of Spinal Muscular Atrophy.

36. (Once Amended) A method for detecting Arthrogryposis Multiplex Congenita (AMC), said method comprising:

(a) extracting DNA from a patient sample;

(b) amplifying said DNA via a polymerase chain reaction (PCR) using unlabeled primers from exon 7 or exon 8 of the Survival Motor Neuron (SMN) gene of SEQ ID No:21;

(c) subjecting said amplified DNA to a Single Stranded Conformation Polymorphism (SSCP); and

(d) detecting the presence or absence of Arthrogryposis Multiplex Congenita.

40. (Once Amended) A method of detecting the presence in a human patient of an altered Survival Motor Neuron (SMN) gene associated with Spinal Muscular Atrophy, comprising:

analyzing exon 7 or exon 8 of a gene identified as T-BCD541 (SEQ ID No: 21) in a biological sample derived from the patient, and

comparing said exon 7 or exon 8 to the corresponding exon of SEQ ID No:13, which is present in a normal tissue;

wherein an alteration of either exon 7 or exon 8 in said patient sample with reference to said normal tissue is indicative of the presence of an altered Survival Motor Neuron (SMN) gene associated with Spinal Muscular Atrophy in said patient.

50. (Once Amended) A method of confirming a clinical diagnosis of Arthrogryposis Multiplex Congenita in a patient, comprising

analyzing exon 7 or exon 8 of a gene identified as T-BCD541 (SEQ ID No : 21) in a biological sample derived from the patient, and

comparing said exon 7 or exon 8 to the corresponding exon of SEQ ID No:13, which is present in a normal tissue;

wherein an alteration of either exon 7 or exon 8 in said patient sample with reference to said normal tissue is indicative of the presence of an altered Survival Motor Neuron (SMN) gene associated with Arthrogryposis Multiplex Congenita in said patient.

Please add the following claims :

53. (New) A kit for the *in vitro* detection of a defect in the Survival Motor Neuron (SMN) gene, wherein said kit comprises a probe which comprises at least 9 nucleotides within a sequence of SEQ ID No: 21 or hybridizes under stringent conditions with a sequence of SEQ ID Nos: 1, 2, 10-13, or 21.

54. (New) A method of identifying the presence or absence of a mutation in the Survival Motor Neuron (SMN) gene in a subject, comprising

(a) isolating a nucleic acid from the subject;

(b) subjecting the nucleic acid to digestion by a restriction endonuclease, wherein restriction fragments resulting from said digestion of a mutated SMN gene differ from those obtained from a T-BCD541 gene of SEQ ID No:21; and

(c) identifying the presence or absence of a mutation in the SMN gene in the subject.

55. (New) The method of claim 54, wherein the restriction endonuclease is *Bsr-1*.

56. (New) The method of claim 54, wherein the nucleic acid is further subjected to a polymerase chain reaction (PCR) following isolation.

57. (New) The method of claim 56, wherein said polymerase chain reaction is performed with a set of primers which are contained in the sequence comprising nucleotides 921 to 1469 of SEQ ID No: 12, or which comprise a sequence selected from SEQ ID Nos: 5 to 8 and 24 to 57.

58. (New) A method of identifying the presence of Spinal Muscular Atrophy (SMA) in a subject, said method comprising:

(a) isolating a nucleic acid from a subject; and

(b) identifying a mutation in a T-BCD541 gene (SEQ ID No: 21);

wherein the presence of a mutation in the T-BCD541 gene is indicative of the presence of SMA in said subject.

59. (New) The method of claim 58, wherein the mutation is a deletion in the T-BCD541 gene (SEQ ID No: 21).

60. (New) The method of claim 59, wherein the deletion comprises a deletion of the entire T-BCD541 gene (SEQ ID No: 21).

61. (New) The method of claim 59, wherein the mutation results in a truncation of the protein product encoded by SEQ ID No: 12.

62. (New) The method of claim 58, wherein the sequence of the isolated nucleic acid is determined by direct sequencing.

63. (New) The method of claim 58, wherein the nucleic acid is further subjected to a polymerase chain reaction (PCR) following isolation.

64. (New) A kit for the *in vitro* detection of a defect in the survival motor neuron gene, comprising:
a set of primers wherein at least one of said primers comprises a sequence selected from SEQ ID Nos: 5 to 8 and 24 to 57;
reagents for an amplification reaction; and
a probe for the detection of the amplified product.

65. (New) A method for detecting a defect in the Survival Motor Neuron gene, said method comprising:

(a) extracting DNA from a patient sample;
(b) amplifying said DNA with primers, wherein at least one of said primers comprises a sequence selected from SEQ ID Nos: 5 to 8 and 24 to 57;
(c) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP); and
(d) detecting the presence or absence of said defect in the Survival Motor Neuron gene, wherein the presence of said defect is indicative of a Survival Motor Neuron disorder.